

16p11.2 GENETIC FOUNDATION



Bringing Hope for the 16p11.2 Community

MEET THE 16P11.2 DEVELOPMENT & COMMUNITY OUTREACH LEAD



*Jennifer Mack
Development & Community Outreach
Lead of 16p11.2 Genetic Foundation*



TENACITY + THERAPY = TUBA + TRIBE

WHEN THE ANSWERS WOULDN'T COME

Jennifer had an uneventful first pregnancy with son Xander who made it all the way to 40 weeks.

Then there was meconium in the amniotic fluid, and some of it made its way into his lungs. She knew of other babies who had gone through something similar and gone home the next day.

Xander stayed for 16 days.

Even then, he left the hospital with traces still in his lungs. Not long after, he was back in the hospital and needed steroid treatment while struggling to coordinate eating and breathing. He was diagnosed with failure to thrive, something that feels almost surreal to Jennifer now, given the very different metabolic and weight challenges he would face later.

Then came pyloric stenosis.

Xander was spitting up everything he took in, at every feeding. When Jennifer would call with concerns, she was told he was probably fine and that she was just a first-time mom.

Twenty-four hours later, she held him as he cried on his third day without food. He was six weeks old and waiting for surgery.

Later, when his development and speech looked different from those of other children his age, she heard another version of the same dismissal: "Boys talk late."

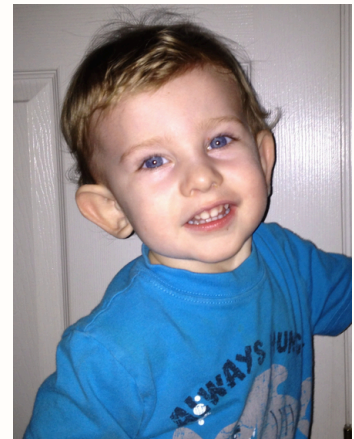
She kept asking questions anyway.

THE DIAGNOSIS CAME EARLY. UNDERSTANDING TOOK LONGER.

Xander received his 16p11.2 deletion diagnosis at age 3.

Over the years, he accumulated more than 25 diagnoses and significant clinical findings. Specialists looked at individual pieces, but Jennifer was still trying to understand the whole child.

She learned having a diagnosis and understanding a diagnosis are two very different things.



*Xander & Jennifer
at the 2025 16p11.2
Conference*

Finding the 16p11.2 Genetic Foundation changed that for her.

She attended the conference in October 2025, and by the end of it, she felt a renewed sense of purpose. She could see the vision Dr. Faranak "Franki" Herrera and the team were building, and she knew she wanted to be part of it.

Franki could take what researchers were learning about 16p11.2, translate it into language that families could understand, and thereby help Jennifer see how that science might apply to the child in front of her.

Suddenly, she could recognize patterns and ask better questions.

She had a clearer picture of why certain things might be happening and how to advocate for Xander as a whole person, rather than as a collection of separate diagnoses.

Franki also changed the way she thought about intervention.

Over the years, Xander has participated in speech therapy, occupational therapy, physical therapy, myofunctional therapy, brain training, and more. As he got older, some of those supports began to trail off. It became easier to accept that school-based speech services could end or that OT might not identify a clear enough goal, especially when Xander was content spending much of his free time on a screen and had no interest in becoming a basketball star.

Franki challenged Jennifer not to let adolescence become the point when they quietly backed away.

Her explanation helped Jennifer understand something she had not fully appreciated before: Xander can absolutely keep learning and making progress, but acquiring new skills may take more repetition and effort for him. Meanwhile, the world around him keeps getting more complicated. His peers are building new academic, social, communication, motor, and independence skills at a rapid pace.

Instead of assuming Xander was done with therapies, Jennifer started asking where he might need more support.

That was one of the most practical gifts of understanding the science better. It changed what she was willing to accept as “good enough” for Xander and reminded her that being content does not mean he has reached his potential.

Families need research. They also need people who can help translate it into: “Here’s what this might mean for your child, and here’s what you may want to think about next.”



TENACITY IN ACTION

Jennifer often thinks about the distance between the newborn who spent weeks struggling to breathe and clear a lung infection and the teenager who now marches onto a football field carrying a tuba, an instrument that depends on breath control.

That still moves her.

Adaptation has characterized Xander’s whole life.

When people could not understand him, he found creative ways to reframe what he was trying to say. Before he could speak at all, that same determination showed up in the way he would climb, reach, and problem-solve his way to whatever he wanted.

One way or another, he kept going.

He has spent his life learning, adapting, practicing, and trying again. Jennifer sees some of her own determination reflected in him.

FINDING A PLACE TO PUT IT TO USE

Jennifer now volunteers roughly 40 hours a week for the 16p11.2 Genetic Foundation while raising four children with significant medical and developmental needs.

She does it because Xander is proof that showing up, over and over, changes things.



This community gave her somewhere to put all those years of searching, questioning, learning, and advocating. Now, **as Director of Development and Community Outreach, she uses those skills to help families find their footing, tell community stories, build awareness, develop partnerships, strengthen donor relationships, and invite people to invest in the mission.**

She also gets to dream up ways to bring people in.

The [Sweet 16 Club](#), the Foundation's monthly giving community, and **16p11.2 Awareness Day on November 16** are two initiatives she is especially proud to have helped start. You'll find out more about both below.

And she sees so much more the Foundation could do.

A VISION BIGGER THAN OUR RESOURCES

There are more families to reach, more stories to tell, more awareness to build, more partnerships to pursue, and more ways the Foundation could support this community with the resources to do it.

That is where help is needed.

The Foundation needs donors and monthly supporters. It needs partners and volunteers. It needs people who will share its work, make an introduction, open a door, or decide that this rare community is worth investing in.

Jennifer knows what it feels like to have a diagnosis in hand and still be trying to understand the child in front of her. She also knows what it feels like when someone finally helps the pieces make more sense.

She wants that understanding to be easier to find for the families coming after hers.

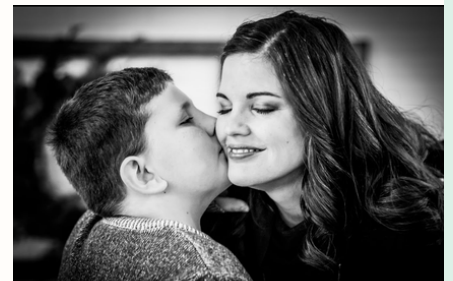
Every person who joins the Foundation gives it a little more capacity to make that happen.

WHAT XANDER HAS TAUGHT JENNIFER

Xander has spent his life working extraordinarily hard to become who he is.

From the baby who struggled to breathe and eat to the teenager carrying a tuba onto a football field, his life has been an exercise in tenacity.

He inspires his mother to bring that same tenacity to what comes next for this community.



[**VOLUNTEER**](#)



[**SUBSCRIBE**](#)



[**DONATE**](#)



JOIN THE SWEET 16 CLUB

Less than two coffees a month. A real difference for kids like Xander.

Children like Xander need a stronger connection between research and clinical care. **The Sweet 16 Club helps the Foundation turn scientific knowledge into guidance that families and clinicians can use.**

For \$16 a month, supporters can join this monthly giving community. Members currently receive perks at least once a month, beginning with a private welcome message from the Foundation's founder, Dr. Franki Herrera. Other benefits include exclusive invitations, early access to opportunities, member discounts, and more. As membership grows, the Foundation hopes to offer perks every week.



BECOME A 16P11.2 AWARENESS DAY AMBASSADOR

November 16. Every state, every country.
Goal: \$16,000.

This year, the Foundation is working to bring 16p11.2 Awareness Day to every U.S. state and also beyond its borders. **We are recruiting national and international ambassadors to lead awareness efforts in their communities.** The fundraising goal is \$16,000, and the Foundation hopes to secure a matching gift to help reach it.

What it means to be an ambassador:

- Commit to donating or raising \$1,600 within your community over the year.
- Promote 16p11.2 awareness and advocacy across your state or country.
- Help connect local families to the Foundation and to one another.

For anyone looking for a meaningful way to give back to families like theirs, becoming an ambassador is a place to start.



**NOVEMBER
16, 2026**

**CELEBRATE
16P11.2 DAY:**



Every year on November 16, our community comes together to recognize 16p11.2 Day, a day to raise awareness, celebrate our incredible families, and support the work of the 16p11.2 Genetic Foundation.

This year, we are inviting every family to join our **Fill My 16 Challenge:**

Find 16 people to donate \$16 each.

That is **\$256 raised by one family!** When our entire community participates, those small donations can make an incredible collective impact.

Funds raised help the 16p11.2 Genetic Foundation support:

- Research and clinical advancement
- Education and trusted resources
- Family and community support
- Better standards of care
- Awareness and advocacy



FUNDRAISING FOR BEGINNERS: IT'S EASIER THAN YOU THINK!

You do not need fundraising experience, a special event, or a huge social media following. All you need is your story, your personalized flyer, and a willingness to ask others to support a cause that matters to your family.

1

CREATE YOUR FUNDRAISING PAGE

Visit our [16p11.2 Day Zeffy fundraising page](#).

Follow the prompts to create your personal peer-to-peer fundraising page. Add your name, a family photo, and a short explanation of why the 16p11.2 Genetic Foundation is important to you.

Your message does not need to be long. Simply share from the heart!

[CLICK HERE](#)



2

PERSONALIZE YOUR “FILL MY 16” FLYER

Use our [customizable Canva template](#) to create your own flyer.

Add your family's photo in the space provided and personalize the wording if you would like. Download your finished flyer as an image so you can easily share it by text, email, or social media.

[CLICK HERE](#)

3

ASK 16 PEOPLE

Think of 16 people who may be willing to help:

- Family members
- Social media friends
- Neighbors
- Friends
- Church members
- Coworkers
- Local businesses
- Teachers and therapists

Send them your flyer and personal fundraising link. You can also post it on Facebook or Instagram.

Remember: **You are not asking one person for \$256. You are asking 16 people for \$16 each.**



4

FILL IN YOUR 16 SPOTS

Each time someone donates \$16, use our [customizable Canva template](#) to fill in or mark off one of the numbered circles on your flyer. Share updates so your friends and family can see your progress.

5

SAY THANK YOU

Thank every person who donates, shares your fundraiser, or helps spread awareness. A short message can make your supporters feel connected to the difference they are helping create.

Together, we can fill every spot, raise awareness, support families, and create a brighter future for everyone affected by 16p11.2.



CONNECTING EDUCATION, CARE, AND RESEARCH

16p WEBINAR SERIES



UPCOMING TOPICS:

- **Call to Action: 16p11.2 Advocacy Session** Hosted by Dr. Faranak Herrera
October 1, 2026 at 11:00am PST
- **16p11.2 Duplication: What We Know and What We Need to Learn**
Hosted by Dr. Peter Kannu
- **16p11.2 Deletion: What We Know and What We Need to Learn**
Hosted by Dr. Faranak Herrera
- **16p11.2 Duplication: Possible Features and Lifelong Trajectories**
Hosted by Dr. Peter Kannu
- **16p11.2 Deletion: Possible Features and Lifelong Trajectories**
Hosted by Dr. Faranak Herrera

Education gives families the knowledge and confidence to advocate for their loved ones, explore research opportunities, and connect with specialized care. Through our Webinar Series and future educational resources, families can become better informed while strengthening connections across the 16p11.2 community.

MISSED ANY OF OUR
PAST WEBINARS?
CATCH UP HERE!



<https://www.youtube.com/@16p11.2GeneticFoundation>

ADVANCEMENTS IN RESEARCH AND CLINICAL CARE FOR 16P11.2 GENETIC SYNDROMES



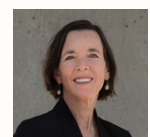
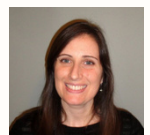
UNIVERSITY of WASHINGTON



Hawk-IDDRC
Intellectual and Developmental
Disabilities Research Center

REGISTER NOW

- **October 13, 2026 6:00 PM EST**
Hosted by Dr. Carly Demopoulos, PhD from University of California San Francisco, Topic: Speech
- **November 10, 2026 6:00 PM EST**
Hosted by Dr. Emily Neuhaus, PhD from University of Washington, Topic: Family Research Priorities
- **December 1, 2026 6:00 PM EST**
Hosted by Dr. Dara S. Manoach, PhD from Lurie Center for Autism, Topic: Sleep



BRINGING THE 16P11.2 COMMUNITY'S VOICE TO GLOBAL GENES

In September 2026, **Dr. Faranak "Franki" Herrera, founder of the 16p11.2 Genetic Foundation, attended the Global Genes Rare Drug Development Symposium in Boston.** She returned with new connections, answers to important questions, and a clearer sense of the work needed to advance research for people with 16p11.2 variants.

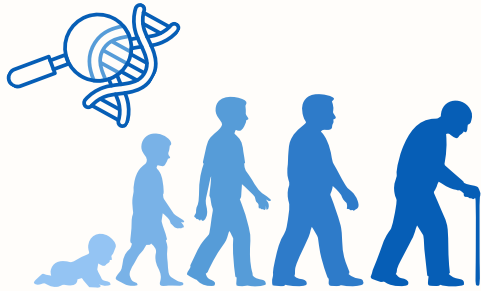


At the symposium, Franki learned from patient advocacy groups further along in their research journeys, including the FOxG1 Research Foundation. She explored how families, researchers, industry, and advocacy organizations can work together to prepare for potential clinical trials. Discussions covered the practical and regulatory challenges of rare disease drug development, ways to bring the patient's voice into that process, and approaches to gene therapy.

The experience reinforced a central priority for Franki: families must help shape the research that affects them. The Foundation made connections with people in the drug development field and will continue advocating for the needs of the 16p11.2 community as it explores possible paths forward.



One topic stood out in particular: natural history studies. These studies follow people over time to help researchers understand how a condition affects them at different ages. That information can reveal patterns, help researchers decide what to measure in a clinical trial, and provide a basis for evaluating whether an intervention makes a difference.



For families, participating may mean completing surveys without seeing an immediate benefit. Franki wants the community to understand why those contributions matter. **Each family's experience adds to the knowledge researchers need to understand 16p11.2 and plan future studies.**

Simons Searchlight collects natural history information for all four 16p11.2 deletion and duplication variants. Families with an eligible, confirmed genetic diagnosis can **register to participate directly.**

SIMONS
SEARCHLIGHT

Driven by science. United by hope.

Franki's trip was an opportunity to learn from others who have navigated the path toward rare disease research and to make sure 16p11.2 families are part of the conversation. The Foundation will bring those lessons and connections into its work to build a stronger foundation for future research.

Dr. Wendy Chung spoke at the Global Genes reception hosted by Simons Searchlight for patient advocacy group leaders. Dr. Chung gathered feedback on how to make research data collection more accessible to their communities.

